Mowat-Wilson Syndrome

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Mowat-Wilson syndrome is a genetic condition that affects many parts of the body. Major signs of this disorder frequently include distinctive facial features, intellectual disability, delayed development, an intestinal disorder called Hirschsprung disease, and other birth defects.

genetic conditions

1. Introduction

Children with Mowat-Wilson syndrome have a square-shaped face with deep-set, widely spaced eyes. They also have a broad nasal bridge with a rounded nasal tip; a prominent and pointed chin; large, flaring eyebrows; and uplifted earlobes with a dimple in the middle. These facial features become more distinctive with age, and adults with Mowat-Wilson syndrome have an elongated face with heavy eyebrows and a pronounced chin and jaw. Affected people tend to have a smiling, open-mouthed expression, and they typically have friendly and happy personalities.

Mowat-Wilson syndrome is often associated with an unusually small head (microcephaly), structural brain abnormalities, and intellectual disability ranging from moderate to severe. Speech is absent or severely impaired, and affected people may learn to speak only a few words. Many people with this condition can understand others' speech, however, and some use sign language to communicate. If speech develops, it is delayed until mid-childhood or later. Children with Mowat-Wilson syndrome also have delayed development of motor skills such as sitting, standing, and walking.

More than half of people with Mowat-Wilson syndrome are born with an intestinal disorder called Hirschsprung disease that causes severe constipation, intestinal blockage, and enlargement of the colon. Chronic constipation also occurs frequently in people with Mowat-Wilson syndrome who have not been diagnosed with Hirschsprung disease.

Other features of Mowat-Wilson syndrome include short stature, seizures, heart defects, and abnormalities of the urinary tract and genitalia. Less commonly, this condition also affects the eyes, teeth, hands, and skin coloring (pigmentation). Although many different medical issues have been associated with Mowat-Wilson syndrome, not every individual with this condition has all of these features.

2. Frequency

The prevalence of Mowat-Wilson syndrome is unknown. More than 200 people with this condition have been reported in the medical literature.

3. Causes

Mutations in the *ZEB2* gene cause Mowat-Wilson syndrome. The *ZEB2* gene provides instructions for making a protein that plays a critical role in the formation of many organs and tissues before birth. This protein is a transcription factor, which means that it attaches (binds) to specific regions of DNA and helps control the activity of particular genes. Researchers believe that the ZEB2 protein is involved in the development of tissues that give rise to the nervous system, digestive tract, facial features, heart, and other organs.

Mowat-Wilson syndrome almost always results from a loss of one working copy of the *ZEB2* gene in each cell. In some cases, the entire gene is deleted. In other cases, mutations within the gene lead to the production of an abnormally short, nonfunctional version of the ZEB2 protein. A shortage of this protein disrupts the normal development of many organs and tissues, which causes the varied signs and symptoms of Mowat-Wilson syndrome.

3.1. The Gene Associated with Mowat-Wilson Syndrome

- ZEB2

4. Inheritance

This condition is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder.

5. Other Names for This Condition

- Hirschsprung disease-mental retardation syndrome
- microcephaly, mental retardation, and distinct facial features, with or without Hirschsprung disease
- MWS

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